Case Report

Sturge-Weber syndrome: A case report

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Received – 09 February 2015 Initial Review – 28 March 2015 Published Online – 23 July 2015

Abstract

Sturge-Weber syndrome (SWS) is a rare disorder that occurs sporadically with a frequency of 1:50,000. It is a neurocutaneous syndrome, characterized by a facial capillary malformation (port-wine stain), abnormal blood vessels on the brain (leptomeningeal angioma), and the eyes leading to glaucoma. The patient may present with seizure, hemiparesis, stroke-like episodes, headache, and developmental delay. Here, we report a case of SWS in a 5-month-old baby presenting with the features of port-wine stain, focal seizure, and intracranial calcification.

Key words: *Sturge-Weber syndrome, Port-wine stain, Intracranial calcification, Leptomeningeal angioma*

Sturge-Weber syndrome (SWS) is a rare disorder that occurs with an incidence of 1:50,000 [1]. The syndrome results from the anomalous development of embryonic vascular bed in the early stages of face and central nervous system (CNS) development [2]. The recent hypotheses, aberrant sympathetic innervations, increased vascular growth factors and defects in the extracellular matrix, are yet to be tested [2]. It is a sporadic neurocutaneous syndrome characterized by facial port-wine stain, eye abnormalities (glaucoma and choroidal hemangioma), and leptomeningeal angioma involving occipital and posterior parietal lobes [3]. The features of this syndrome are facial nevus, seizures, hemiparesis, intracranial calcification, and mental retardation [2].

Angiomas involve the leptomeninges and skin of the face typically in ophthalmic and maxillary distributions of the trigeminal nerve [4]. Developmental disorders are more common when angiomas are bilateral [5].

CASE REPORT

A 5-month-old infant born to non-consanguinous parents was brought to the emergency department with left sided focal seizure with facial twitching of 20 min duration associated with frothing from mouth and loss of consciousness. She was born full term following uneventful pregnancy and delivery. She was the third child of her parents, and her two siblings were apparently normal. The child was able to roll over and hold things given to her, and she was able to babble and laugh aloud. She was fully immunized for her age.

Her weight, length, and head circumference were 6.7 kg (within −1 standard deviation [SD] and median), 64 cm (median), and 42 cm (within the median and 1 SD), respectively. Her physical examination revealed that she was afebrile with a heart rate of 120/min and respiratory rate of 30/min. The port-wine stains were present over upper right half of the face and eyelids (Fig. 1). The oral cavity had no port-wine stain and other abnormalities. Her neurological and physical examinations were found to be normal. Ophthalmological examination revealed no abnormality and intraocular pressure was 15 mmHg and 13 mmHg in right and left eye, respectively.

Hematological and the biochemical profile were within normal limits. Skull radiograph was normal. Computed tomography (CT) scan of the brain showed intracranial leptomeningeal calcification in the subcortical area of the right parietal lobe (Fig. 2). The patient was advised for the magnetic resonance (MR) angiography brain for further assessment of the intracranial lesion but the parents refused.

After admission to the pediatric ward, peripheral venous access was established. Appropriate anticonvulsants, antibiotics, and fluids were administered. Her seizure was controlled after anticonvulsant therapy with phenytoin, and there was no recurrence. There was no postictal neurological deficit. She was discharged on 5th day of admission with anticonvulsant therapy and was kept on regular follow-up. Feature suggestive of SWS in this child were port-wine stain, focal seizures opposite to the side of nevus and intracranial calcification.

DISCUSSION

SWS is a rare neurocutaneous disorder, which is classified into three types on the basis of organ involvement by Roach scale. In Type I, both face and leptomeningeal angiomas are present, and there may be glaucoma; in Type II, only facial

Figure 1: Port-wine stain in right upper part of face

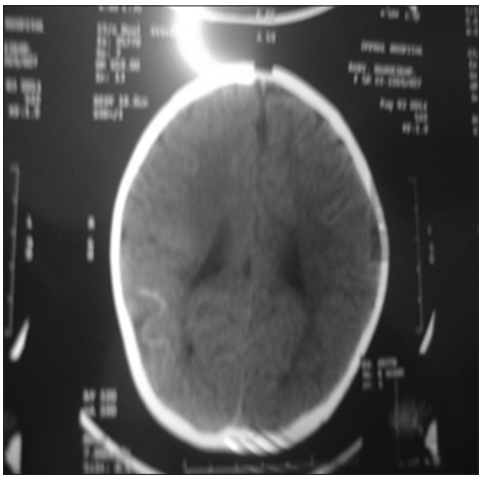

Figure 2: Computed tomography scan of brain showing leptomeningeal calcification in right parietal lobe

angiomas are seen, and there may be glaucoma; and Type III has isolated leptomeningeal angiomas and no glaucoma [2]. Our patient had Type I SWS as the patient had both CNS and facial involvement, but MR angiographic imaging would have given more details whether the leptomeningeal calcification is an angioma [2].The incidence of SWS is sporadic [4]. Both the sexes are equally affected, and no racial difference has been reported [5]. Although the precise pathogenesis is unknown, it is thought to be the result of the anomalous development of the primordial vascular bed during the early stages of cerebral vascularization [2].

Its diagnosis is established by port-wine stain in the face accompanied by other signs such as glaucoma, epilepsy, developmental delay, mental retardation and skull radiograph, CT, and MR imaging (MRI) scans [2]. Indocyanine green angiography can provide information that is not detected by clinical or fluorescence angiographic examination in patients with SWS. CT scan brain of our patient showed intracranial calcification in the right parietal region. We could not do indocyanine green angiography due to lack of diagnostic facilities. This may be important and sensitive in detecting the diffuse choroidal hemangioma associated with SWS [6]. The complications can be stroke-like episodes, glaucoma, developmental delay, and behavior abnormalities [2]. Hence, regular monitoring of intraocular pressure is important [2].

Treatment involves early control of seizures and prevention of complications [4]. Pulsed dye laser therapy for port-wine stains can be done for the cosmetic purpose [2]. Hemispherectomy and other epilepsy surgeries can be useful in refractory seizures and for localized epileptic focus in one hemisphere [2]. The parents of all the diagnosed patients must receive counseling regarding the potential risk of complications.

SWS must be differentiated from Klippel–Trenaunay–Weber syndrome, which also has hypertrophy of bone and soft tissue along with the clinical manifestations of SWS [7]. Beckwith–Wiedemann syndrome and Dyke-Davidoff-Masson syndrome also have to be differentiated [7]. Siderosis also results in atrophy of one cerebral hemisphere that is similar to that seen in SWS. Imaging studies (MRI with contrast) that outline cerebral vasculature will be necessary to differentiate between these disorders [7].

CONCLUSION

SWS should be suspected in any child presenting with port-wine stain over the face and seizure. All the suspected cases should be thoroughly examined and investigated to confirm the diagnosis.

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Funding: None; Conflict of Interest: None Stated.

How to cite this article: De D, Periyakaruppan M, Prakash SC, Sunilbala K, Singh KI, Singh LR. Sturge-Weber syndrome: A case report. Indian J Child Health. 2015;2(3):145-146.